POSTER CO182

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ePROVIDE Searches for Rare Disease Functioning and Epilepsy Measures in Children with KCNQ2.

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Background

KCNQ2-DEE is a rare, pediatric condition that manifests as several significant functional and developmental impairments. Disease-specific clinical outcome assessment (COA) instruments evaluate treatment benefits in the KCNQ2-DEE population and inform drug development, however no validated COAs have been developed for KCNQ2-DEE clinical trial applications at this time.

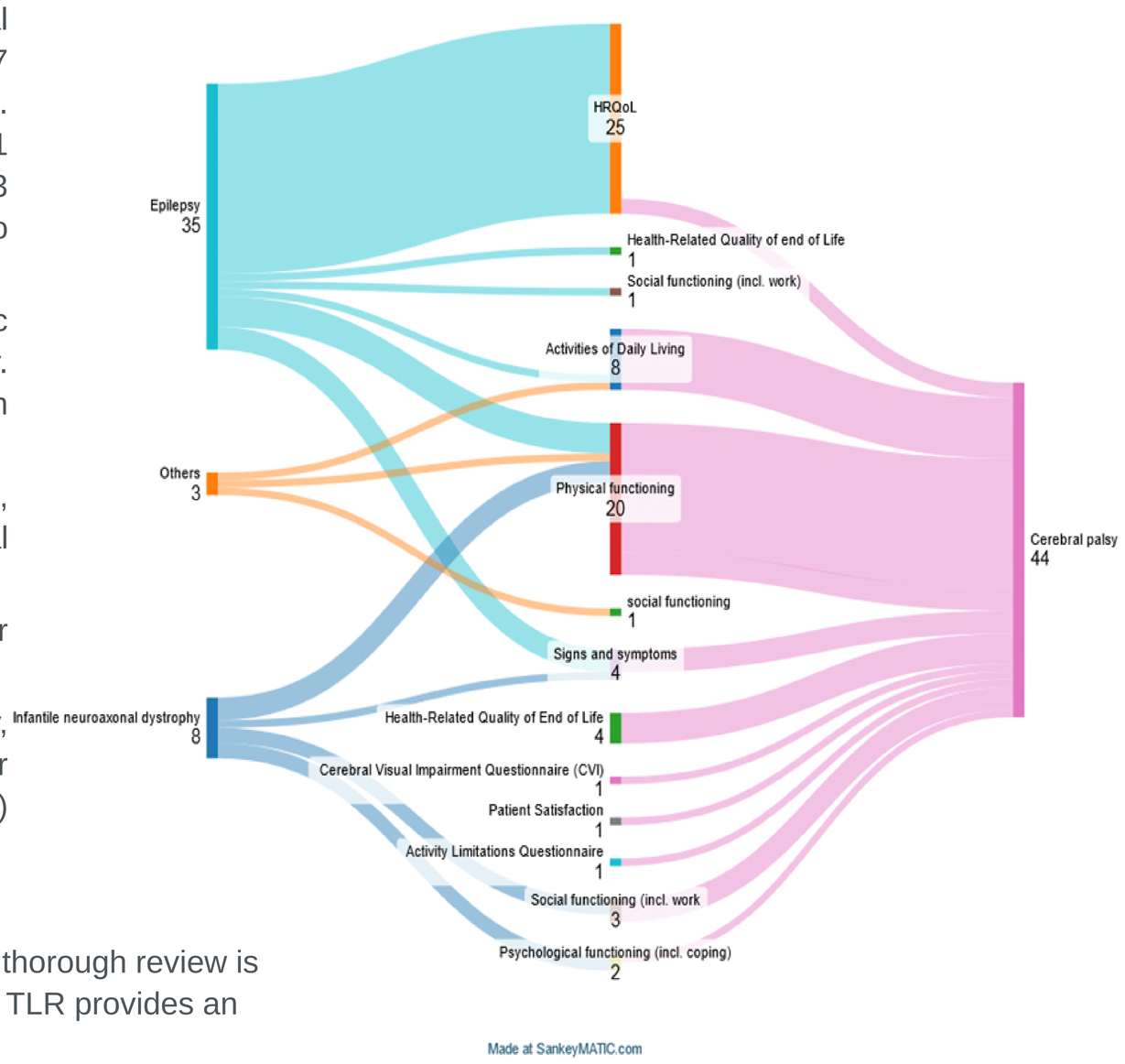
Methods

A search was conducted on the ePROVIDE PROQOLID[1] database to identify and summarize the characteristics of existing COA instruments relevant to children with KCNQ2-DEE. Characteristics evaluated included domains and items evaluated by the scale, respondent type (patient, caregiver, clinician, or observer), response scaling, and disease-specific or generic measures to explore potential suitability for use in clinical trials.

Results

- A total of 78 COAs were identified in the PROQOLID[1] database from three relevant disease areas associated with KCNQ2-DEE: Epilepsy, Cerebral Palsy (CP), and Infantile Neuroaxonal Dystrophy (IAD). For Epilepsy, 17 PROs, 12 ObsROs, 1 ClinRO, and 2 Performance tests were identified. For CP, 8 PROs, 20 ObsROs, 6 ClinROs, 2 Performance tests, and 1 composite instrument was identified. For IAD, the database included 3 PRO and 6 ObsROs. All instruments were reviewed for suitability to measure treatment effects and disease progression in KCNQ2-DEE.
- Concepts of importance to outcomes researchers or health economic benefit ratings (e.g., the ability to walk, and dress) were measured rarely. In contrast, complex emotions, such as engagement or communication with parents are less frequently captured.
- Although numerous PROs were identified in the PROQOLID[1] database, they are not appropriate for severely disabled children who are nonverbal and of low reading age.
- ObsROs were identified in each category and were the source of further evaluation.
- As some KCNQ2-DEE children suffer multiple seizures occurring daily, starting within the first week of life, of interest could be for older children(Glasgow Epilepsy Outcome Scale for Young Persons (GEOS-YP) and Epilepsy and Learning Disabilities Quality of Life Scale (ELDQOL).

Figure 2 Sankey Diagram Showing Concepts of Interest related to three major disease areas covered by PROQOLID databases



Summary

Although many scales exist for clinical diseases similar to KCNQ2-DEE, a thorough review is needed to identify the right items to create a new KCNQ2-DEE scale. This TLR provides an indication of relevant scales that cover content. The Sankey diagram

Figure 1: The treemap represents various Scales that were identified within Age groups of respective Diseases

Epilepsy Scales for Different Age Groups

Childhood (1 to 11 years)	Adolescent (12-17 years)	Infancy (0-1 year of age)
(PedsQL™ Epilepsy Module), (GEOS-YP),(ELDQOL,(QOLCE), (QOLCE-16), (ICIS), (Neuro-QoL Short Form v1.1 - Pediatric Depression), (Neuro-QoL Scale v1.1 - Pediatric Lower Extremity), (Neuro-QoL Short Form v2.0 - Pediatric Cognitive Function), (Neuro-QoL Scale v1.1 - Pediatric Upper Extremity - Fine Motor, ADL), (Neuro-QoL Item Bank v1.0 - Pediatric Social Relationships - Interaction With Peers), (Neuro-QoL Item Bank v1.0 - Pediatric Stigma), (Neuro-QoL Short Form v1.0 - Pediatric Anger), (Neuro-QoL Short Form v2.1 - Pediatric Fatigue), (Neuro-QoL Short Form v1.0 - Pediatric Pain), (Neuro-QoL Short Form v1.0 - Pediatric Social Relationships - Interaction with Peers), (Pediatric Neuro-QOL SF), (DISABKIDS Smiley version - SR version), (QOLCE-55)	(PedsQL™ Epilepsy Module), (GEOS-YP), (ELDQOL), (QOLCE), (DISABKIDS EM - Proxy), (CHEQOL), (QOLCE-16), (QOLIE-AD-48), (ICIS), (Neuro-QoL Short Form v1.1 - Pediatric Depression), (Neuro-QoL Scale v1.1 - Pediatric Lower Extremity), (Neuro-QoL Short Form v2.0 - Pediatric Cognitive Function), (Neuro-QoL Scale v1.1 - Pediatric Upper Extremity - Fine Motor, ADL), (Pediatric Neuro-QOL SF), (DISABKIDS DCGM-37 - Proxy), (DISABKIDS Smiley version - SR version), (QOLCE-55), (RLIES)	(PedsQL™ Epilepsy Module), (ELDQOL)(DISABKIDS EM - Proxy),(CHEQOL),(DISABKIDS DCGM-37 - Proxy) **Adult (18 and above)** (PedsQL™ Epilepsy Module), (ELDQOL), (RLIES)

Cerebral Palsy Scales for Different Age Groups

Adolescent (12-17 years)	Childhood (1 to 11 years)	Infancy (0-1 year of age)
(DIS),(PedsQL™ 3.0 Cerebral Palsy Module),(DISABKIDS CPM - Proxy), (DISABKIDS CPM - SR version), Functional Mobility Scale (FMS), (ChARM), (FAQ), (GMFM-66-IS), (ABILHAND - KIDS), (ABILOCO-Kids), (PODCI-Parent), (ASK©-Capability), (ASK©-Performance), (DHI), (COSA), (DISABKIDS EM - Proxy), (DISABKIDS EM - SR version), (RSBQ)	(DIS), (PedsQL™ 3.0 Cerebral Palsy Module), (ChARM), (FAQ), (PMAL), (GMFM-66), (GMFM-66-B&C), (ABILHAND - KIDS), (ABILOCO-Kids), (CEDL v2), (CEDL or Child Engage), (ASK©-Capability), (ASK©-Performance), (EDVA), (DHI), (RSBQ)	(DISABKIDS CPM - Proxy), (DISABKIDS CPM - SR version), Functional Mobility Scale (FMS), (GMFM-66), (C-BiLLT), (GMFM-66-IS), (CEDL v2), (PMAL-R), (CEDL or Child Engage), (PODCI-Parent), (EDVA), (COSA), (DISABKIDS EM - Proxy), (DISABKIDS EM - SR version), (N-PASS) **Adult (18 and above)** Functional Mobility Scale (FMS), (FAQ), ,(PODCI-Parent), (DHI)

Infantile Neuroaxonal Dystrophy Scales for Different Age Groups

Adolescent (12-17 years)	Adult (18 and above)	Infancy (0-1 year of age)
(PedsQL™ Duchenne Muscular Dystrophy Module), (DMD-QoL), (DMD Impact Measure), (ABILHAND – Neuromuscular Disorders), (UEFI-15)	(PedsQL™ Duchenne Muscular Dystrophy Module), (DMD-QoL), (DMD Impact Measure), (UEFI-15) **Childhood (1 to 11 years)** (PedsQL™ Duchenne Muscular Dystrophy Module), (ABILHAND – Neuromuscular Disorders)	(DMD-QoL),(DMD Impact Measure), (DMD CaGI-C)

Conclusions

- No single COA identified in the review appeared to have robust content validity for assessing treatment benefits in KCNQ2-DEE across all age groups.
- An indication-specific tool is needed to be developed that provides comprehensive and patient-centric outcomes measurement for KCNQ2-DEE patients and their caregivers.

REFERENCES

[1] Caron M, Perrier LL, Vaissier V, Savre I, Acquadro C, Emory MP. MAPI Research Trust, Lyon, France.